

IMMUNOASSAYS TO DETECT DISEASES OR DISEASE SUSCEPTIBILITY TRAITS

ABSTRACT OF THE DISCLOSURE

- 5 Disclosed are immunoassay methods for the diagnosis/prognosis of diseases and disease susceptibility traits associated with gene mutations that cause protein truncation or allelic loss. The levels of one or more targeted wild-type proteins expressed by a subject gene or genes are immunologically quantitated in biological samples. Results indicating that a targeted wild-type protein is not present in an
- 10 assayed sample, or that approximately 50% of the normal amount of such a wild-type protein is present in an assayed sample are considered to be positive for a mutation in one or both alleles of a subject gene, and correlated with the disease or the disease susceptibility trait associated with that mutation or mutations. Normal cells, particularly normal peripheral blood lymphocytes, are preferred biological samples.